Disease Name — Phenylketonuria

Alternate name(s)	Hyperphenylalaninemia, Phenylalanine hydroxylase deficiency, Følling disease
Acronym	PKU
Disease Classification	Amino Acid Disorder
Variants	Yes
Variant name	Benign phenylketonuria, Mild phenylketonuria, Variant phenylketonuria, Biopterin-responsive phenylketonuria Tetrahydrobiopterin deficiencies: GTP cyclohydrolase I deficiency, 6-Pyruvoyl-tetrahydropterin synthase deficiency, Dihydropteridine reductase deficiency, Pterin-4_-carbinolamine dehydratase deficiency
Symptom onset	Infancy
Symptoms	Mental retardation, decreased pigmentation relative to family members, eczematous rash, seizures, abnormal gait, and unusual “mousy” odor to urine.
Natural history without treatment	Mental retardation in the moderate to severe range, hyperactivity, eczema, mild neurologic manifestations, possible abnormal gait microcephaly.
Natural history with treatment	If diet instituted early, normal IQ and development can be expected.
Treatment	Dietary restriction of phenylalanine with supplementary formula for tyrosine and essential amino acids.
Other	“Mousy” or “musky” smelling urine. Females with PKU are at-risk to have children affected by maternal PKU (increased levels of phenylalanine are teratogenic).
Emergency Medical Treatment	See sheet from American College of Medical Genetics (attached) or for more information, go to website: http://www.acmg.net/StaticContent/ACT/Phenylalanine.pdf
Physical phenotype	No abnormalities present at birth. May develop widely-spaced incisors, pes planus, epicanthus and microcephaly.
Inheritance	Autosomal recessive
General population incidence	1:10,000
Ethnic differences	Yes
Population	Turks, Irish
Ethnic incidence	Turks (1:2600), Irish (1:4500)
Enzyme location	Liver
Enzyme Function	Converts phenylalanine to tyrosine
Missing Enzyme	Phenylalanine hydroxylase
Metabolite changes	Increased plasma phenylalanine, increased phenylpyruvic acid in urine, decreased plasma tyrosine.
Prenatal testing	DNA testing is possible if mutations known. RFLP analysis is successful in 75% of families.
MS/MS Profile	N/A
OMIM Link	http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=261600
Genetests Link	www.geneclinics.org
Support Group	National Urea Cycle Disorders Foundation http://www.nucdf.org National Coalition for PKU and Allied Disorders http://www.pku-allieddisorders.org/ Children Living with Inherited Metabolic Diseases http://www.climb.org.uk/

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